AMERICAN JOURNAL OF PHYSICAL ANTHROPOLOGY 101:145–149 (1996)

Torus Mandibularis in 45,X Females (Turner Syndrome)

LASSI ALVESALO, JOHN T. MAYHALL, AND JUHA VARRELA
Institute of Dentistry, University of Oulu. FIN-90220 Oulu, Finland (L.A.): Faculty of Dentistry, University of Toronto, Toronto M5G 1G6, Canada (J.T.M., J.V.)

KEY WORDS Chromosome aneuploidy, Sex chromosomes, Torus mandibularis, Human

ABSTRACT Ninety-three Finnish females with a 45,X chromosome constitution, 78 first-degree female, and 37 first-degree male relatives were examined to determine the frequency and expression of torus mandibularis. The results indicate that among adults the frequency of the trait was significantly lower and the expression weaker in the 45,X females than in male control relatives. A similar trend was observed in comparison to normal females. In juveniles the trend was reversed. Our findings suggest that the sex chromosomes may have an influence on the occurrence, expression, and timing of development of the mandibular torus. Sexual dimorphism in the manifestation of torus mandibularis may result particularly from the effect of the Y chromosome on growth.

Torus mandibularis is a bony prominence or exostosis found on the lingual surface of the mandibular corpus, above the mylohyoid line. It is usually bilateral and located in the premolar segment. Most of the hypotheses put forward to explain the development of mandibular tori emphasize either genetic or nongenetic influence (Eggen, 1989; Ossenberg, 1981). Several early workers suggested that the presence of tori is influenced by functional factors (Hooton, 1918; Hrdlička, 1940; Kajava, 1912; Schreiner, 1935; van der Broek, 1943). The effect of masticatory activity was demonstrated by Mayhall and Mayhall (1971), who studied the occurrence of the tori in prehistoric and modern Inuit populations. They found that the frequency of the tori in Inuit, who used a traditional diet, was approximately the same as in their ancestors, whereas in those who subsisted on a European diet tori were less common. Subsequent studies have further clarified the role of functions related to mastication in the development of the tori (Eggen and Natvig, 1986; Hylander, 1977; Ossenberg, 1981).

Some comparative studies have shown that there are population differences in the manifestation of torus mandibularis which do not seem to depend on diet or other environmental factors (Cambell, 1925; Lasker, 1950; Mayhall et al., 1970; Moorrees, 1957; Moorrees et al., 1952). A genetic component in the development of tori has been confirmed by family studies (Alvesalo and Kari, 1972; Johnson et al., 1965; Krahl, 1949; Suzuki and Sakai, 1960). Johnson et al. (1965) found that the torus is inherited as an autosomal dominant trait, whereas Alvesalo and Kari (1972) concluded that the mode of inheritance is autosomal recessive. However, as Ossenberg (1981) has pointed out, none of the models explains the pedigree data satisfactorily. The most recent findings (Eggen, 1989) suggest that about 30% of the variation in torus mandibularis is genetically determined, whereas 70% can be attributed to an environmental influence.

Sex differences in the occurrence of man-

Received July 31, 1995; accepted April 4, 1996.

Address reprint requests to Dr. Lassi Alvesalo, University of Oulu, Institute of Dentistry, Aapistie 3, FIN-90220 Oulu, Finland.

dibular tori have also been found. For example, torus frequency among the men of the population of Hailuoto was almost twice that of the women. Such differential distribution might occur if sex chromosome genes modify the manifestation of the mandibular torus (Alvesalo and Kari, 1972).

We have studied the occurrence of torus mandibularis in 45,X females and in their normal female (46,XX) and male (46,XY) first-degree relatives. We seek to determine whether the sex chromosomes moderate the development of mandibular tori, and thereby test Alvesalo and Kari's (1972) hypothesis.

SUBJECTS AND METHODS

Ninety-three Finnish females with a 45,X chromosome constitution, 78 first-degree female relatives (46,XX), and 37 first-degree male relatives (46,XY) were examined to determine the frequency and expression of torus mandibularis. The groups were divided into juveniles (under 17 years of age) and adults, and these were analyzed separately. The mean ages of juvenile and adult 45,X females were 12.45 years (SD 3.37) and 23.45 years (SD 6.45), respectively; those of control females were 12.67 years (SD 3.48) and 33.60 years (SD 10.24), respectively, and those of control males were 11.07 years (SD 3.93) and 37.65 years (SD 10.41), respectively.

The assessments of the sizes of the tori were made by palpation during clinical oral examinations by the same examiner (L.A.). Eggens' (1954) scale was slightly modified to rate the expression as absent, small, medium, large, and extra large. In our statistical analyses large and extra large were combined. Both unilateral and bilateral expressions were considered as positive, with the larger being recorded in the case of a bilateral expression. Chi-square and, when frequencies were too small, Fisher's exact test were used to evaluate the presence or absence of tori between 45,X females and their 46,XX or 46,XY relatives. Finally, the same statistics were used to assess differences between 46,XX females and 46,XY males.

RESULTS

Table 1 indicates that in adult 45,X females the raw frequency of mandibular torus is less than it is in either their male or female relatives. The raw frequency in control males is higher than in the control relatives. Because of the small number of individuals with torus expression, statistical testing was confined to the analysis of the presence or absence of the tori. The probabilities derived from the statistical tests (Table 2) indicate that in adults the difference between 45,X females and their first-degree male relatives is significant ($P < 0.05$). In the other comparisons, the differences are not significant. However, inspection of Table 3 indicates that trends are apparent. If only those over 16 years of age are considered and all categories of expression are combined, the frequency of torus mandibularis in the 45,X females, in the female controls, and in the male controls increases with each sex chromosome constitution. Such an increase in expression over X, XX, and XY also occurs when medium and large tori are combined. Finally, the magnitude of expression of small tori relative to all other tori is given by the ratio of small tori to medium and large tori combined. The ratio is largest in 45,X females, is smaller in males, and is lowest in normal females (Table 3).

In the juveniles (under 17 years of age) it was not possible to determine the ratio of small to medium and large tori (Table 1). However, a trend in expression could be observed. The trend is reversed in juveniles

TABLE 1. Frequency of torus mandibularis by age and expression

Age (years)	≤16		>16		Total	
Expression	N	%	N	%	N	%
45,X females						
None	32	80.0	42	79.2	74	79.6
Small	1	2.5	4	7.5	5	5.4
Medium	7	17.5	7	13.2	14	15.1
Large	0	0.0	0	0.0	0	0.0
Total	40	100.0	53	100.0	93	100.0
Male relatives						
None	13	92.2	13	56.5	26	70.3
Small	0	0.0	2	8.7	2	5.4
Medium	1	7.1	7	30.4	8	21.6
Large	0	0.0	1	4.3	1	2.7
Total	14	100.0	23	100.0	37	100.0
Female relatives						
None	13	86.7	47	74.6	60	76.9
Small	0	0.0	2	3.2	2	2.6
Medium	2	13.3	13	20.6	15	19.2
Large	0	0.0	1	1.6	1	1.3
Total	15	100.0	63	100.0	78	100.0

TABLE 2. Presence[1] absence of torus mandibularis in 45,X females and first-degree male and female relatives

	45,X females vs. normal males			Normal males vs. females			Normal females vs. 45,X females		
	45,X females Present/ total	%	*P*	Normal males Present/ total	%	*P*	Normal females Present/ total	%	*P*
0–16 years	8/40	20.0	0.254	1.14	7.1	0.526	2.15	13.3	0.445
Adults	11/53	20.8	0.042	10/23	43.5	0.106	16/63	25.4	0.556

[1] Presence category = small, medium, large, and extra large expressions combined. Probabilities derived from Chi-square and Fisher's exact tests.

compared to adults. 45,X females show the highest frequency of torus mandibularis (20.0%), control males show the lowest (7.1%), and control females are in between (13.3%).

DISCUSSION

Our results indicate that the overall frequency of torus mandibularis is significantly lower in adult 45,X females than in their male relatives. The frequency difference with their normal female relatives was in the same direction, but was not significant. The degree of expression of the torus in 45,X females also appears weaker than in the controls. The mean age of the adult 45,X females is lower than that of the control groups, a factor which may have affected the results if the development of the torus in 45,X females continues in adulthood. However, our findings suggest that this may not be the case. The frequency of torus mandibularis in the juvenile 45,X females is similar to that of adult 45,X females with only a negligible increase from the age of 12 years to 23 years. This suggests that the development of the mandibular tori in 45,X females may take place earlier than in normal individuals. This is of interest because it has been shown that tooth formation and dental eruption are advanced in 45,X females compared to normal females (Filipson et al., 1965; Kari and Alvesalo, 1985). The growth of the tori would, therefore, be in pace with the dental development rather than with growth of the facial or postcranial skeleton (Filipson et al., 1965; Park et al., 1983; Tanner et al., 1959).

The 45,X females are considerably shorter than normal females (Brook et al., 1974; Varrela et al., 1984). It might therefore be argued that the weaker and less frequent appearance of torus mandibularis in 45,X females could be related to their smaller stature, or their diminished rate of growth in comparison with 46,XX females. With the exception of the cranial base, the cranial growth of the 45,X females seems to be normal and in most cranial dimensions the 45,X females do not differ from normal females (Peltomäki et al., 1989; Varrela, 1984). The mandible of the 45,X females seems to be slightly shorter but, at the same time, broader than in normal 46,XX females (Laine and Alvesalo, 1986). However, even if a positive correlation between torus size and stature is established, indicating that small individuals tend to have small expressions of the tori, this may not influence the validity of our deductions, at least, with regard to the presence or absence of the tori. Indeed, if the assessment is biased by a general size factor, the same bias must also be considered in the comparisons between normal males and females.

The 45,X females show an increased frequency of occlusal anomalies (Laine et al., 1986). As masticatory stress is a factor which may modify the expression of the tori (Mayhall and Mayhall, 1971), it seems possible that the occlusal discrepancies might lower the level of masticatory activity and decrease stimulation of the bone growth. On the other hand, it seems unlikely that dietary factors could explain the present findings since

TABLE 3. Expression of torus mandibularis in adults (age >16 years)

Torus expression	Sex chromosome constitution: 45,X female	XX female	XY male
Present[1]	20.8%	25.4%	43.5%
Combined[2]	13.2%	22.2%	30.4%
Small/combined	0.57	0.14	0.25

[1] Small + medium + large tori.
[2] Combined = medium + large tori.

there are few, if any, differences in diet and social conditions between the 45,X females and their relatives (Takala et al., 1985). On similar grounds it seems unlikely that other influences related to diet which have been suggested to affect the development of the tori, e.g., mucosal irritation, would have any bearing on the present findings.

Hormonal findings in 45,X females include low estrogen and androgen levels and increased secretion of gonadotropins; growth hormone levels in 45,X females seem to be within normal limits and thyroid function is usually normal (Simpson, 1976). There is little evidence to indicate that any of the hormonal changes would directly affect the growth of the 45,X females, except in the development of secondary sex characteristics (Simpson, 1976) and pelvic growth at puberty (Varrela et al., 1984). It thus seems unlikely that a decrease in the frequency and expression of the torus mandibularis in 45,X females would be caused by hormonal alterations. Normally, low levels of the sex hormones tend to retard maturation. Therefore, early development of the tori would indicate that their growth is independent of action of the sex hormones.

Dental development is affected by genes on the X and Y chromosomes (Alvesalo, 1985). An enamel gene has been located on the X chromosome, which, unlike the Y chromosome, has very little or no influence on the growth of the dentine. The Y chromosome promotes growth of both enamel and dentine, possibly through a regulative action (Alvesalo, 1985; Alvesalo et al., 1985, 1987, 1991, Alvesalo and Tammisalo, 1981). The Y chromosome may influence dentine growth by an effect on cell proliferation resulting in larger average tooth crown sizes in males compared to females. Tooth crown size in 45,X females is smaller than in normal females (Alvesalo and Tammisalo, 1981; Filipson et al., 1965; Kari et al., 1980; Town send et al., 1984). Thus, the effect of a "missing" X or Y chromosome on growth of the mandibular tori seems to be analogous to its decreasing effect on tooth size. The variation in enamel thickness apparently results from prolonged matrix secretion of the ameloblasts, whereas the tori grow by periosteal apposition which depends on both cell proliferation and matrix secretion. Therefore, a sexual dimorphism in the frequency of torus mandibularis, as suggested by our results together with some earlier finding (Alvesalo and Kari, 1972; Hakkila, 1944; Moorrees et al., 1952), could be explained by the different action of the X and Y chromosomes on the assumption that their effect on growth is pleiotropic. The effect of the Y chromosome on cell proliferation would be particularly important in this respect.

Our findings indicate that, compared to normal males, both the total frequency and expression of torus mandibularis in 45,X females are decreased. In addition, the development of torus mandibularis seems to take place earlier in 45,X females than in normal males or females. This result can be explained by the involvement of human sex chromosomes in the regulation of growth of the tori. We suggest that sexual dimorphism in the manifestation of torus mandibularis may be a consequence of the effect of the Y chromosome on growth.

ACKNOWLEDGMENTS

This study was supported by the University of Turku Foundation and the Academy of Finland. We thank the anonymous reviewers and the editor for their helpful suggestions.

LITERATURE CITED

Alvesalo L (1985) Dental growth in 47,XYY males and in conditions with other sex-chromosome anomalies. In AA Sandberg (ed.): The Y Chromosome, Part B: Clinical Aspects of the Y Chromosome Abnormalities. London: Alan R. Liss, pp. 277–300.

Alvesalo L, and Kari M (1972) Hailuodon hampaistotutkimus V. Torus mandibularis: Esiintyminen ja periytymiseen liittyviä näkökohtia. Proc. Finn. Dent. Soc. *68:*307–314.

Alvesalo L, and Tammisalo E (1981) Enamel thickness in 45,X females' permanent teeth. Am. J. Hum. Genet. *33:*355–359.

Alvesalo L, Tammisalo E, and Therman E (1987) 47,XXX females, sex-chromosomes and tooth crown structure. Hum. Genet. *77:*345–348.

Alvesalo L, Tammisalo E, and Townsend G (1991) Upper central incisor and canine tooth crown size in 47,XXY males. J. Dent. Res. *70*1057–1060.

Brook CGD, Mürset G, Zahlman M, and Prader A (1974) Growth in children with 45,XO Turner's syndrome. Arch. Dis. Child. *49:*789–795.

Cambell TD (1925) Dentition and Palate of the Australian Aborginal. Adelaide: Hassell.

Eggen S (1954) Torus mandibularis og marginale paradentopatier. Odontol. Tidskr. *62:*431–442.

Eggen S (1989) Torus mandibularis: An estimation of the degree of genetic determination. Acta. Odont. Scand. *47:*409–413.

Eggen S, and Natvig B (1986) Relationship between torus mandibularis and number of present teeth. Scan. J. Dent. Res. *94:*233–240.

Filipson R, Lindsten J, and Almqvist S (1965) Time of eruption of the permanent teeth, cephalometric and tooth measurement and sulphation factor activity in 45 patients with Turner's syndrome with different types of X-chromosome aberrations. Acta Endocrinol. *48:*91–113.

Hakkila S (1944) Ylä-ja alaleuan toruksista. Suom. Hammaslääk. Toim. *LXXIX:*9–34.

Hooton EA (1918) On certain Eskimoid characters in Icelandic skulls. Am. J. Phys. Anthropol. *1:*53–76.

Hrdlička A (1940) Mandibular and maxillary hyperostoses. Am. J. Phys. Anthropol. *27:*1–55.

Hylander WL (1977) The adaptive significance of Eskimo craniofacial morphology. In: AA Dahlberg and TM Graber (eds.): Orofacial Growth and Development. The Hague: Mouton, pp. 301–330.

Johnson CC, Gorlin RJ, and Anderson VE (1965) Torus mandibularis: A genetic study. Am. J. Hum. Genet. *17:*433–439.

Kajava Y (1912) Die Zähne der Lappen. Suom. Hammaslääk. Toim. *X:*5–66.

Kari M, and Alvesalo L (1985) Dental maturity in 45,X females. J. Dent. Res. *64:*342.

Kari M, Alvesalo L, and Manninen K (1980) Sizes of deciduous teeth in 45,X females. J. Dent. Res. *59:* 1383–1385.

Krahl VE (1949) A familial study of palatine and mandibular tori. Anat. Rec. *103:*447.

Laine T, Alvesalo L, Savolainen A, and Lammi S (1986) Occlusal morphology in 45,X females. J. Craniofacial Genet. Dev. Biol. *6:*351–355.

Lasker GW (1950) Genetic analysis of racial traits of the teeth. Cold Spring Harbor Symp. Quant. Biol. *XV:*191–203.

Mayhall JT, and Mayhall MF (1971) Torus mandibularis in two Northwest Territories Villages. Am. J. Phys. Anthropol. *34:*143–148.

Mayhall JT, Dahlberg AA, and Owen DG (1970) Torus mandibularis in Alaskan Eskimo population. Am. J. Phys. Anthropol. *33:*57–60.

Moorrees CFA (1957) The Aleut Dentition. Cambridge: Harvard University Press.

Moorrees CFA, Osborne RH, and Wilde E (1952) Torus mandibularis: Its occurrence in Aleut children and its genetic determinants. Am. J. Phys. Anthropol. *10:* 319–329.

Ossenberg NS (1981) Mandibular torus: A synthesis of new and previously reported data and a discussion of its cause. In JS Cybulski (ed.): Contribution to Physical Anthropology, 1978–1980. Ottawa: National Museum of Canada, pp. 1–52.

Park E, Bailey JD, and Cowell CA (1983) Growth and maturation in patients with Turner's syndrome. Pediatr. Res. *17:*1–7.

Peltomäki T, Alvesalo L, and Isotupa K (1989) Shape of the craniofacial complex in 45,X females: Cephalometric study. J. Craniofacial Genet. Dev. Biol. *9:*331–338.

Schreiner KE (1935) Zur Osteologie der Lappen. Inst. Sammenlignende Kulturforskning Ser. B *18:*161–177.

Simpson JL (1976) Disorders of Sexual Differentiation. London: Academic Press.

Suzuki M, and Sakai T (1960) A familial study of torus palatinus and torus mandibularis. Am. J. Phys. Anthropol. *18:*263–272.

Takala I, Alvesalo L, Palin-Palokas T, Paunio K, and Suoranta K (1985) Caries prevalence in Turner's syndrome (45,X females). J. Dent. Res. *64:*126–128.

Tanner JM, Prader A, Habich H, and Ferguson-Smith MA (1959) Genes on the Y chromosome influencing rate of maturation in man. Skeletal age studies in children with Klinefelter's (XXY) and Turner's (XO) syndromes. Lancet *2:*141–144.

Townsend G, Jensen BL, and Alvesalo L (1985) Reduced tooth size in 45,X (Turner syndrome) females. Am. J. Phys. Anthropol. *64:*367–372.

van der Broek AJP (1943) On exostoses in the human skull. Acta Neerland. Morphol. *5:*95–118.

Varrela J (1984) Effects of the X and Y chromosome on body size and shape. Proc. Finn. Dent. Soc. *80:* Suppl. V, 1–59.

Varrela J, Vinkka H, and Alvesalo L (1984) The phenotype of 45,X females: An anthropometric quantificatin. Ann. Hum. Biol. *11:*53–66.